Case Report

Congenital Erythropoietin Porphyria (CEP) – A Case of Necrotic Scleritis

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Purpose: To discuss a case of bilateral temporal sclera necrosis leading to scleral perforation.

Material and Methods: This is an observational case report in an institutional setting, reporting an unusual case. A 26 years old young man, farmer by occupation presented to Nishtar Hospital Multan with pain and photophobia in his left eye. Examination showed bilateral involvement of temporal sclera leading to sclera perforation. The visual acuity was 20/20 on log Mar in both eyes. The pupils were reactive to light and Intra-ocular pressure was 14mmHg and 12 mmHg in right and left eye respectively. Dilated fundus examination was found to be normal with Cup – Disc ratio of 0.3 both eyes. Investigation (CBC, peripheral blood film, CRP, Rh factor, X-ray chest, biochemical analysis of specimen) led to unusual case of Gunther's disease also known as congenital erythropoietin porphyria 3.

Congenital erythropoietic porphyria (CEP) is very rare metabolic disorder affecting the synthesis of haem, the iron-containing pigment that binds oxygen onto red blood cells. Cutaneous signs of Gunther's disease (congenital erythropoietic porphyria) may develop 5 years after the onset of symptomatic thrombocytopenia[1]. Other symptoms include alopecia, hypertrichosis, and cutaneous blistering disease involving face, neck, and dorsum of hands, deformities of cartilages of ear and nose etc[2].

Occular signs of this disease may develop from as first decade to 3rd decade of life[3]. Ocular symptoms include pain, photophobia and lacrimation while signs include congestion, scleritis, sclera necrosis and ulceration sometimes leading to uveal prolapsed as well.

MATERIAL AND METHODS

This is an observational case report carried out at Nishtar Hospital Multan. A young male of 26 years age presented to out patient department with pain and photophobia in left eye and grittiness in his right eye. His symptoms had existed for 15 days back. He had a history of blistering skin disease affecting his face, neck, dorsum of hands and causing disfigurement of his nose, ear and proximal digits of upper limbs. He was farmer by occupation. One of his cousins had similar complaints in childhood but had now recovered except few cutaneous lesions. On examination it revealed acute scleritis with scleral perforation almost 1cm in diameter just 2 mm away from the limbus. In the base of ulcer ciliary body some yellowish deposits was visible at the margins of the ulcer. Two telengiectatic vessels were present in juxta ulcer area with corneal haze in juxta limbal area. The visual acuity was 20/20 on log Mar in both eyes. The pupils were reactive to light and Intra-Ocular pressure was 14mmHg and 12 mmHg in right and left eye respectively. Dilated fundus examination was found to be normal with cup-disc ratio of 0.3 both eyes. Systemic evaluation revealed no abnormality and complete blood cell count was within normal range with low hemoglobin level was low while ESR and CRP was normal. No lesion was seen on x-ray chest PAV. Rheumatoid factor was found to be negative.

He was admitted in the ward and mild topical steroids (Florometholone e/d QID) along with oral steroids (Tab. Prednisolone 5 mg, 3 / PO / QID) and

topical lubricants (Tears Plus e/d QID) were started. After 3 days of treatment symptoms were improved but photophobia was still severe. The fascia lata was grafted on sclera on both sides to fill the gap and strengthen the sclera. During surgery hard crystals were removed from the wound margin[4], which after laboratory examination showed calcium and porphyrin. On right side wound margins were quite healthy but on left side melting of sclera was present. After grafting, conjunctival flap was transposed. After 48 hours the right eye was quite normal and left eye showed congestion. Patient was discharged on topical and oral steroids and topical lubricants. On follow up after 7 days, his wounds were quite normal and asymptomatic.

DISCUSSION

CEP is an inherited disorder in which there is a mutation in a gene on chromosome 10 that encodes uroporphyrinogen III synthase. It is transmitted in autosomal recessive pattern. Carriers of a single abnormal gene do not usually exhibit any sign or symptom of the disorder. Due to deficiency of uroporphyrinogen III synthase, Hydroxymethylbilale is not converted into uroporphyrinogen III instead it is converted into uroporphyrinogen I and caproporphyrinogen I which accumulates in the body as uroporphyrin I and caproporphyrin I respectively[5]. This disease affects primarily the Red Blood Cells leading to increased number of immature RBCs in the blood. Very rarely it affects the sclera causing necrotic scleritis, but can cause loss vision[6,7]. Treatment involves oral steroids, rapid blood transfusion for anemia and splenectomy for thrombocytopenia.

CEP if associated only with scleritis is treated with topical and oral steroids and intensive prevention of exposure to sunlight by wearing dark goggles containing UV filters and topical lubricants. Advanced cases with scleral perforations require surgical interventions like facia lata graft.

CONCLUSION

In cases of spontaneous scleral necrosis especially when bilateral rare disease like porphyria should not be missed. In this case as serological investigation did not show any significant evidence, localized toxic response causing local inflammation was probably the culprit.

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